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OPIS PRZYPADKU
CASE REPORT

Unusual presentation of small bowel obstruction in Cowden syndrome: A case report and literature review

Nietypowy obraz kliniczny niedrożności jelita cienkiego w zespole Cowden – opis przypadku i przegląd piśmiennictwa

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ABSTRACT

Intestinal intussusception in adults is a rare disorder, occurring in about 5% of cases. In the paediatric population, it is most often idiopathic in origin, while in adults it is associated with pathologies within the intestinal wall. The purpose of this study is to emphasise the need for expanded diagnosis in adult patients with symptoms of gastrointestinal obstruction due to intestinal intussusception. The medical records of a 22-year-old female patient hospitalised in the Department of General Surgery and Coloproctology of St Alexander Hospital in Kielce, Poland were analysed. The patient presented to the hospital with complaints of abdominal pain accompanied by vomiting. She had a history of treatment for Hashimoto's disease, iron deficiency anaemia and gastritis. After the initial diagnosis, the patient was transferred to the department of general surgery, where conservative treatment was initiated and gastroscopy was performed. During hospitalisation, the patient was consulted in internal medicine due to uncompensated hypothyroidism. Due to a lack of improvement, imaging studies were ordered, which revealed a dilated loop of the small intestine with suspected intussusception. The patient was qualified for surgery, during which an intussusception of the jejunum was found. Due to the inability to drain the intussusception, a segmental small bowel resection was performed. Histopathological examination revealed a hamartomatous polyp as the cause of the intussusception. Given the overall clinical picture, the patient was referred to the genetic counselling centre, where Cowden syndrome was confirmed. Intestinal intussusception, although rare, should be considered as a potential cause of gastrointestinal obstruction in adults. Identifying the pathological changes that cause it is crucial to the overall diagnostic and therapeutic process.

KEYWORDS

Cowden syndrome, hamartomatous polyp, intussusception

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STRESZCZENIE

Wgłobienie jelita u osób dorosłych jest schorzeniem rzadkim, stanowiącym około 5% wszystkich przypadków. W populacji pediatrycznej najczęściej ma charakter idiopatyczny, natomiast u dorosłych wiąże się z patologiami w obrębie ściany jelita. Celem niniejszej pracy jest podkreślenie konieczności rozszerzenia diagnostyki u dorosłych pacjentów z objawami niedrożności przewodu pokarmowego spowodowanej wgłobieniem jelita. Przeanalizowano dokumentację medyczną 22-letniej pacjentki hospitalizowanej na Oddziale Chirurgii Ogólnej i Koloproktologii Szpitala Kieleckiego św. Aleksandra. Pacjentka zgłosiła się do szpitala z bólem brzucha i wymiotami. W przeszłości była leczona z powodu choroby Hashimoto, niedokrwistości z niedoboru żelaza oraz zapalenia błony śluzowej żołądka. Po wstępnej diagnozie pacjentkę przekazano na oddział chirurgii ogólnej, gdzie wdrożono leczenie zachowawcze i wykonano gastroskopię. Podczas hospitalizacji pacjentkę skierowano na konsultację na oddział chorób wewnętrznych z powodu niewyrównanej niedoczynności tarczycy. Ze względu na brak poprawy stanu zdrowia zlecono badania obrazowe, które wykazały rozszerzenie pętli jelita cienkiego z podejrzeniem wgłobienia. Pacjentkę zakwalifikowano do operacji, podczas której stwierdzono wgłobienie jelita czczego. Ze względu na brak możliwości odprowadzenia wgłobienia wykonano segmentową resekcję jelita cienkiego. Badanie histopatologiczne wykazało, że przyczyną wgłobienia był polip hamartomatyczny. Ze względu na ogólny obraz kliniczny pacjentkę skierowano do poradni genetycznej, gdzie potwierdzono zespół Cowden. Wgłobienie jelita, choć rzadkie, należy brać pod uwagę jako przyczynę niedrożności przewodu pokarmowego u osób dorosłych. Identyfikacja zmian patologicznych będących jego przyczyną ma kluczowe znaczenie dla całego procesu diagnostyczno-terapeutycznego.

SŁOWA KLUCZOWE

zespół Cowden, polip hamartomatyczny, wgłobienie jelita

INTRODUCTION

Intussusception, a form of intestinal obstruction, is often described as the intussusception of one segment of intestine into an adjacent segment. The vast majority of cases of intestinal intussusception are treated by paediatric surgeons, as the condition is more common in children than in adults. It is estimated that about 5% of all intussusception cases occur in adults [1]. Considering all cases of intestinal obstruction in adults, only about 1% are due to intussusception. In adults, these cases are usually associated with pathological conditions, while in children they are usually idiopathic [2,3]. The most common underlying pathology in adult intussusception is a benign tumour (about 37%). Most of the lesions are located in the intestine [4]. Hamartomatous polyps may be associated with Cowden syndrome (CS) and may pose a risk of intussusception, which can lead to intestinal obstruction [5,6]. CS is a rare genetic disorder caused by mutations in the phosphatase and tensin homolog gene (*PTEN*) that most commonly affects young people in the second and third decades of life [7]. The incidence is about 1 in 200,000, and it is slightly more common in women [8]. Among other things, the disease is characterised by multiple hamartomatous lesions occurring in various organs [9,10]. It is most commonly manifested by cutaneous and mucosal lesions, which include acral keratosis or oral papillomatosis [11,12]. The syndrome leads to an increased risk of malignancies, including thyroid and colorectal cancers [13]. The *PTEN* gene plays a key role in gastrointestinal and cancerous processes, including hamartomatous polyposis syndromes, which are genetic conditions characterised by polyps in the gastrointestinal tract [14]. Individuals with colorectal cancer are often found to have preexisting colorectal polyposis. Most individuals with at least one mutant allele of the *PTEN* gene have polyps in the large intestine (90%–95% of cases) [11]. Polyps also occur in the small intestine, but the risk of tumorigenesis is not specified in the literature [15].

The medical data of a 22-year-old female patient were retrospectively reviewed, including clinical presentation, laboratory and imaging studies and surgical findings. Abdominal computed tomography (CT) scans, gastroscopy and histopathological findings from resected intestinal specimens were analysed. The diagnostic and therapeutic approach was consistent with current clinical guidelines for suspected small bowel obstruction and intussusception. Informed consent for publication was obtained from the patient.

CASE REPORT

A 22-year-old female patient presented to the Emergency Department with complaints of abdominal pain and associated vomiting of gastric contents. Physical examination revealed tenderness to palpation in the mid-epigastric and left mid-abdomen regions, accompanied by increased tension of the abdominal wall. No peritoneal symptoms were found. In addition, the abdomen was slightly bloated, peristalsis was preserved and there were no abnormalities on per rectum examination. The patient had been treated for anaemia and hypothyroidism since the age of 5 years. In addition, she is under gynaecological care due to heavy menstruation. As a result, the patient was taking Euthyrox, iron and ad hoc cycloamine. On admission, her vital signs were normal. The patient reported a pain score of 7/10 on the numerical rating scale (NRS).

In laboratory tests, a case of severe anaemia with a haemoglobin level of just 4.2 g/dL was observed. The mean corpuscular volume was notably low (66.56 fL) on 20 September 2024, and it fluctuated between 76.28 and 80.6 fL during hospitalisation. Additionally, serum iron levels were low (24 µg/dL) on 23 September 2024, dropping to 18 µg/dL the following day (Table I). The patient was transfused with three units of red blood cell concentrate, and conservative treatment was implemented. Given her medical history, the patient was consulted gynaecologically. Due to nonspecific complaints of abdominal pain, a CT scan of the abdomen and pelvis without contrast was performed. Based on the examination, intussusception of the bowel was suspected. The patient was admitted to the department of general surgery, where conservative treatment was continued; the patient was given gastrografin. An internal medicine consultation was required due to uncompensated hypothyroidism. Gastroscopy was also performed, which revealed superficial gastritis. During hospitalisation, the patient developed extrapyramidal symptoms after the administration of metoclopramide, which necessitated a head CT scan. On the second day after admission, a follow-up CT scan of the abdomen and pelvis was performed, which showed free abdominal fluid and a dilated loop of intestine, again raising the suspicion of intestinal intussusception. Considering the results of the follow-up abdominal CT scan and the lack of improvement after conservative treatment, the patient was qualified for urgent surgery. Intraoperatively, distended loops of bowel and a significant amount of fluid in the abdominal cavity were found. Due to the inability to perform the procedure safely by laparoscopy, the decision was taken to convert to laparotomy. After palpation inspection of the abdominal cavity, an intussusception of the jejunum about 5 cm from the ligament of Treitz was found.

Table I. Laboratory findings during hospitalisation

Date	MCV (fL)	Serum iron (µg/dL)
2024 September 20	66.56	–
2024 September 21	76.28–76.55	–
2024 September 22	77.43	–
2024 September 23	78.09	24
2024 September 24	78.9	18
2024 September 26	80.6	–
2024 September 28	80.25	–
2024 September 29	79.64	–
2024 September 30	79.77	–

MCV – mean corpuscular volume

Due to the inability to drain the intussusception, the decision was taken to perform a segmental resection of the jejunum. During the preparation of the bowel segment, spontaneous drainage of the intussusception occurred, thus making visible a microperforation of the jejunum. After resection of the bowel segment an end-to-side anastomosis was performed, and the patency and blood supply of the anastomosis were checked. A Redon drain was deposited into the smaller pelvis, and the wound was sutured in layers. The bowel fragment was submitted for histopathological examination with a suspected proliferative lesion of the jejunum. The postoperative period went without complications. An oral diet was gradually expanded, which the patient tolerated well. She was discharged home on the eighth postoperative day. In view of the overall clinical picture, the patient was referred to the genetic counselling centre, where CS was confirmed.

DISCUSSION

In the United States, the incidence of small bowel obstruction is estimated to be approximately 350,000 cases per year. The most common causes are adhesions (65%), hernias (10%), cancer (5%) and Crohn's disease (5%) [16].

Invagination of the intestine (intussusception) occurs mainly in the paediatric population, where it is most often not accompanied by a discernible structural cause. In adults, this condition is rare – accounting for about 5% or 1% of all intussusception cases – and is characterised by a nonspecific clinical presentation, which significantly complicates accurate diagnosis [1,2,3,17]. Intussusception in adults is often associated with structural lesions, with tumours or polyps accounting for approximately 35% of cases [18]. A meta-analysis of 1,229 patients showed malignancy in 32.9%, benign lesions in 37.4% and idiopathic aetiology in 15.1% of cases [4]. Structural causes were identified in 85.7% of cases, including benign (35.3%) and malignant (64.7%) tumours [17]. Lead points may include fibroid polyps or submucosal lipomas [19,20]. A 22-year-old patient was admitted with intestinal obstruction resulting from intussusception caused by a juvenile hamartomatous polyp. Genetic testing confirmed a *PTEN* mutation. Up to 95% of adults with *PTEN* mutations develop polyps, which can be morphologically diverse, including adenomas, hamartomas, juvenile, lipomatous, leiomyomatous, lymphoid and hyperplastic polyps [21]. According to the National Comprehensive Cancer Network (NCCN) diagnostic criteria, CS/*PTEN* hamartoma tumour syndrome (PHTS) is diagnosed based on the presence of major features (e.g. breast cancer, endometrial cancer, follicular thyroid cancer, macrocephaly, adult Lhermitte–Duclos disease, gastrointestinal hamartomas or multiple characteristic mucocutaneous lesions) and secondary features (e.g. colorectal cancer,

renal cell carcinoma, autism spectrum disorder, lipomas or vascular malformations). A clinical diagnosis is established when an individual meets at least three major criteria (including at least one of the following: macrocephaly, Lhermitte–Duclos disease or gastrointestinal hamartomas) or two major and at least three secondary criteria. In families with a confirmed *PTEN* mutation or a known clinical diagnosis of PHTS, it is sufficient to meet two major criteria, or one major and two minor criteria, or at least three minor criteria [10].

It is worth noting that intussusception in adults is rare, but when it does occur it is often accompanied by a cancerous change, as was the case in our situation. It should also be remembered that in young patients – like our case – the symptoms of CS may be incomplete, which requires special diagnostic vigilance.

CT is the first-line test in the diagnosis of intestinal intussusception. It allows for both the identification of the cause and the planning of appropriate treatment [17]. An X-ray of the abdominal cavity can be helpful as an initial examination in patients with suspected intestinal obstruction. Magnetic resonance imaging offers greater accuracy in assessing blockages, but due to the cost and time required it remains a supplementary method [22]. For patients in stable condition with a history of abdominal surgery, conservative treatment should be considered first. Oral administration of a hypertonic, water-soluble contrast agent may have a therapeutic effect and accelerate the resolution of the obstruction. Research has shown that such treatment reduces the need for surgical intervention and shortens the duration of hospitalisation [22,23]. In case of a worsening clinical condition, urgent surgical intervention is necessary [22]. The choice between laparoscopy and laparotomy depends on the anatomical conditions and the patient's general condition. In case of small intestine obstruction, the laparoscopic technique may be difficult due to the distension of the intestinal loops and the thinning of their walls, which increases the risk of perforation [24]. In clinical practice, a hybrid approach is often used, starting laparoscopically and converting to laparotomy if necessary [25], as in the case at hand. A lower recurrence rate is noted in patients who underwent surgical treatment compared to those treated conservatively for intestinal obstruction. However, the length of hospitalisation was longer among patients treated with surgery [26]. Laparoscopy, compared to laparotomy, is usually associated with a shorter procedure and a shorter hospital stay [27].

This report has some limitations, including the absence of genetic testing in family members, which can provide valuable information about inheritance patterns. Additionally, the short postoperative follow-up period did not allow for an assessment of long-term outcomes or recurrence. Finally, the findings cannot be generalised due to it being a single-patient case study.

CONCLUSIONS

Intestinal intussusception in adults, although rare, can be the first clinical manifestation of CS. This case emphasises the importance of considering genetic syndromes in young adults presenting with nonspecific gastrointestinal symptoms, especially when there are coexistent endocrine and haematologic disorders. Surgical intervention remains the mainstay of treatment in cases of bowel obstruction caused by intussusception, while genetic counselling and lifelong surveillance for associated malignancies are essential in confirmed cases of CS.

Conflict of interest

The authors declare no conflict of interest.

Authors' contribution

Study design – N. Dardzińska, B. Molasy

Data collection – N. Dardzińska, E. Obrębski

Data interpretation – N. Dardzińska, B. Molasy

Manuscript preparation – N. Dardzińska, E. Obrębski, D. Lorek, P. Jaskulska, N. Krzysztofek, K. Jończyk

Literature research – N. Dardzińska, E. Obrębski

Final approval of the version to be published – B. Molasy, N. Dardzińska, E. Obrębski, D. Lorek, P. Jaskulska, N. Krzysztofek, K. Jończyk

REFERENCES

1. Yalamarthi S, Smith RC. Adult intussusception: case reports and review of literature. Postgrad Med J. 2005;81(953):174–177. doi: 10.1136/pgmj.2004.022749.

2. Alshoabi SA, Abdulaal OM. An unusual case of colonic intussusception in old age. J Taibah Univ Med Sci. 2019;14(2):199–202. doi: 10.1016/j.jtumed.2019.02.004.

3. Teyha PS, Chandika A, Kotecha VR. Prolapsed sigmoid intussusception per anus in an elderly man: a case report. J Med Case Rep. 2011;5:389. doi: 10.1186/1752-1947-5-389.

4. Hong KD, Kim J, Ji W, Wexner SD. Adult intussusception: a systematic review and meta-analysis. Tech Coloproctol. 2019;23(4): 315–324. doi: 10.1007/s10151-019-01980-5.

5. McGarrity TJ, Amos CI, Baker MJ. Peutz-Jeghers Syndrome. In: Adam MP, Feldman J, Mirzaa GM, et al. [eds.]. GeneReviews® [Internet]. Seattle (WA): University of Washington; 1993–2025 [updated 2021 Sep 2]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1266/.
6. Gorji L, Albrecht P. Hamartomatous polyps: Diagnosis, surveillance, and management. World J Gastroenterol. 2023;29(8):1304–1314. doi: 10.3748/wjg.v29.i8.1304.
7. Isa HM, Mohamed ZS, Isa ZH, Busehail MY, Alaradi ZA. Cowden syndrome: A rare cause of intestinal polyposis. Cureus. 2024;16(7):e64838. doi: 10.7759/cureus.64838.
8. Manimaran S, Ramanathan R, Subramanian S. Pediatric challenges associated with Cowden syndrome and Graves' disease: A case report. Cureus. 2024;16(4):e58090. doi: 10.7759/cureus.58090.
9. Garofola C, Jamal Z, Gross GP. Cowden Disease. StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Mar 27. Available from: https://www.ncbi.nlm.nih.gov/books/NBK525984/.
10. Takayama T, Muguruma N, Igarashi M, Ohsumi S, Oka S, Kakuta F, et al. Clinical guidelines for diagnosis and management of Cowden syndrome/PTEN hamartoma tumor syndrome in children and adults – secondary publication. J Anus Rectum Colon. 2023;7(4):284–300. doi: 10.23922/jarc.2023-028.
11. Pîrlog LM, Pătrășcanu AA, Militaru MS, Cătană A. Insights into clinical disorders in Cowden syndrome: A comprehensive review. Medicina (Kaunas). 2024;60(5):767. doi: 10.3390/medicina60050767.
12. Lim A, Ngeow J. The skin in Cowden syndrome. Front Med (Lausanne). 2021;8:658842. doi: 10.3389/fmed.2021.658842.
13. Tischkowitz M, Colas C, Pouwels S, Hoogerbrugge N; PHTS Guideline Development Group; European Reference Network GENTURIS. Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. Eur J Hum Genet. 2020;28(10):1387–1393. doi: 10.1038/s41431-020-0651-7.
14. Cotton JL, Dang K, Hu L, Sun Y, Singh A, Rajurkar MS, et al. PTEN and LKB1 are differentially required in Gli1-expressing mesenchymal cells to suppress gastrointestinal polyposis. Cell Rep. 2022;40(3):111125. doi: 10.1016/j.celrep.2022.111125.
15. Vanoli A, Grillo F, Furlan D, Arpa G, Grami O, Guerini C, et al. Small bowel epithelial precursor lesions: A focus on molecular alterations. Int J Mol Sci. 2021;22(9):4388. doi: 10.3390/ijms22094388.
16. Rami Reddy SR, Cappell MS. A systematic review of the clinical presentation, diagnosis, and treatment of small bowel obstruction. Curr Gastroenterol Rep. 2017;19(6):28. doi: 10.1007/s11894-017-0566-9.
17. González-Carreró Sixto C, Baleato-González S, García Palacios JD, Sánchez Bernal S, Junquera Olay S, Bravo González M, et al. Intestinal intussusception in adults: Location, causes, symptoms, and therapeutic management. Radiologia (Engl Ed). 2023;65(3):213–221. doi: 10.1016/j.rxeng.2022.10.005.
18. Su T, He L, Zhou T, Wu M, Guo Y, Wang Q, et al. Most adult intussusceptions are caused by tumors: a single-centre analysis. Cancer Manag Res. 2020;12:10011–10015. doi: 10.2147/CMAR.S268921.
19. Tarchouli M, Ait Ali A. Adult intussusception: An uncommon condition and challenging management. Visc Med. 2021;37(2):120–127. doi: 10.1159/000507380.
20. Boyack I, Vu D, Patel P, Opsha O. Colocolic intussusception secondary to submucosal lipoma. Am J Emerg Med. 2020;38(8):1697.e1–1697.e3. doi: 10.1016/j.ajem.2020.04.046.
21. Pilarski R. *PTEN* hamartoma tumor syndrome: A clinical overview. Cancers (Basel). 2019;11(6):844. doi: 10.3390/cancers11060844.
22. Jackson P, Vigiola Cruz M. Intestinal obstruction: evaluation and management. Am Fam Physician. 2018;98(6):362–367.
23. Jaanimäe L, Lepner U, Kirsimägi Ü, Nikkolo C. Management of small bowel obstruction and therapeutic role of contrast media: A multicenter study. Surg Pract Sci. 2022;12:100149. doi: 10.1016/j.sipas.2022.100149.
24. Quach G, Zielinski MD. Surgical management of small bowel obstruction: What you need to know. J Trauma Acute Care Surg. 2024;96(3):357–363. doi: 10.1097/TA.0000000000004195.
25. Petrou NA, Bonelli EM, Watson N, Wood J, Kontovounisios C, Behar N. Laparoscopic management of acute small bowel obstruction in non-selected patients: A 10-year experience. J Clin Med. 2022;11(21):6275. doi: 10.3390/jcm11216275.
26. Williams SB, Greenspon J, Young HA, Orkin BA. Small bowel obstruction: conservative vs. surgical management. Dis Colon Rectum. 2005;48(6):1140–1146. doi: 10.1007/s10350-004-0882-7.
27. Wang J, Cheng L, Liu J, Zhang B, Wang W, Zhu W, et al. Laparoscopy vs. laparotomy for the management of abdominal trauma: A systematic review and meta-analysis. Front Surg. 2022;9:817134. doi: 10.3389/fsurg.2022.817134.